



## RAB7A gene

RAB7A, member RAS oncogene family

### Normal Function

The *RAB7A* gene provides instructions for making a protein that is involved in endocytosis, a process that brings substances into the cell. During endocytosis, the cell membrane folds around a substance outside the cell (such as a protein) to form a sac-like structure called a vesicle. The vesicle is drawn into the cell and is pinched off from the cell membrane. The RAB7A protein plays a role in vesicle trafficking, which is the movement of these vesicles within the cell.

### Health Conditions Related to Genetic Changes

#### Charcot-Marie-Tooth disease

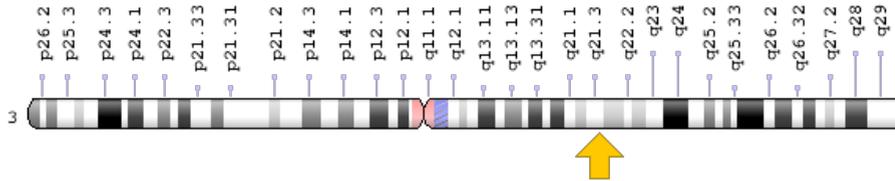
Researchers have identified at least four *RAB7A* gene mutations that cause a form of Charcot-Marie-Tooth disease known as type 2B. This form of the disorder is characterized by a severe reduction of sensation, especially in the limbs. The *RAB7A* gene mutations identified in individuals with this disorder change single protein building blocks (amino acids) used to make the RAB7A protein. These changes probably alter the structure of the RAB7A protein and impair its function.

It is not clear how *RAB7A* gene mutations cause the signs and symptoms of Charcot-Marie-Tooth disease. Researchers suggest that the altered RAB7A protein may affect endocytosis and transport of neurotransmitters, which are chemical messengers that carry signals between nerve cells (neurons). The specialized extensions of neurons (axons) that transmit nerve impulses throughout the nervous system can be very long, especially in the peripheral nervous system. The peripheral nervous system consists of nerves connecting the brain and spinal cord to muscles and sensory cells that detect sensations such as touch, pain, heat, and sound. The extremely long axons of the peripheral nervous system may be particularly sensitive to impaired transport of neurotransmitters. Disrupted neurotransmitter signaling may result in the weakness and sensory problems characteristic of Charcot-Marie-Tooth disease.

## Chromosomal Location

Cytogenetic Location: 3q21.3, which is the long (q) arm of chromosome 3 at position 21.3

Molecular Location: base pairs 128,726,136 to 128,814,798 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- PRO2706
- PSN
- RAB7
- RAB7\_HUMAN
- Ras-associated protein RAB7

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Intracellular Vesicular Traffic  
<https://www.ncbi.nlm.nih.gov/books/NBK21045/>
- Molecular Biology of the Cell (fourth edition, 2002): Rab Proteins Help Ensure the Specificity of Vesicle Docking  
<https://www.ncbi.nlm.nih.gov/books/NBK26859/#A2325>

### GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1285>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28RAB7%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- RAS-ASSOCIATED PROTEIN RAB7  
<http://omim.org/entry/602298>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_RAB7A.html](http://atlasgeneticsoncology.org/Genes/GC_RAB7A.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=RAB7A%5Bgene%5D>
- HGNC Gene Family: RAB, member RAS oncogene GTPases  
<http://www.genenames.org/cgi-bin/genefamilies/set/388>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=9788](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9788)
- Inherited Peripheral Neuropathies Mutation Database  
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=23>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/7879>
- UniProt  
<http://www.uniprot.org/uniprot/P51149>

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